



EBF3-HADDS



International Family Conference

TEXAS CHILDREN'S HOSPITAL

July 25-27, 2019

HOUSTON, TEXAS



Welcome to Houston!

On behalf of our conference planning committee, we would like to welcome you to our first HADDS family conference! We hope this weekend will provide you with new knowledge about HADDS, connect you with other HADDS families, and help you feel energized and hopeful about the exciting developments we have in store for increasing awareness, fundraising, and research for HADDS.

We have 22 families from all over the US and Canada joining us for this conference! We hope you enjoy your time in Houston and explore some of the exciting things the city has to offer, including amazing restaurants, museums, the Houston Zoo, and Hermann park. There is always plenty to do! Please reach out to anyone of us if you have questions or concerns about the conference or exploring Houston--we want to ensure you have a wonderful experience!

Welcome!

Ashley LeMaire, Kelly Mastin, Pilar Magoulas, & Mallory Schindler

Location & Directions

Directions to the conference center

All conference talks on Thursday and Saturday will be held at the Texas Children's Hospital (TCH) Pavilion for Women, Legacy Tower
6651 Main Street, Houston, Texas 77030



It is about a 10 minute walk from the hotel to the conference area. Exit the Hilton lobby and take a left towards Southgate Drive. Take a left at Southgate (first left). Stay on sidewalk and walk towards TCH. Cross Main Street towards TCH. Enter the closest ramp (directly near your left) and walk through the closest TCH entrance (near the Wells Fargo sign, see below—this is important, as there are many TCH entrances). Look for conference signage. Once in the lobby take the elevators on your right to the 4th floor. At the fourth floor, you will see signs above that point to the Conference Center. Head in that direction until you see the registration desk for the HADDs Conference. Please contact Ashley LeMaire (832-331-4166) or Mark LeMaire (228-596-4026) for directions or assistance if you get lost.



Directions to the Neurological Research Institute (NRI)



The NRI is too far to walk in the Houston July heat! Take the Hilton shuttle to the location, which is in the medical center. However, allow yourself plenty of time to catch the shuttle before your scheduled tour time.

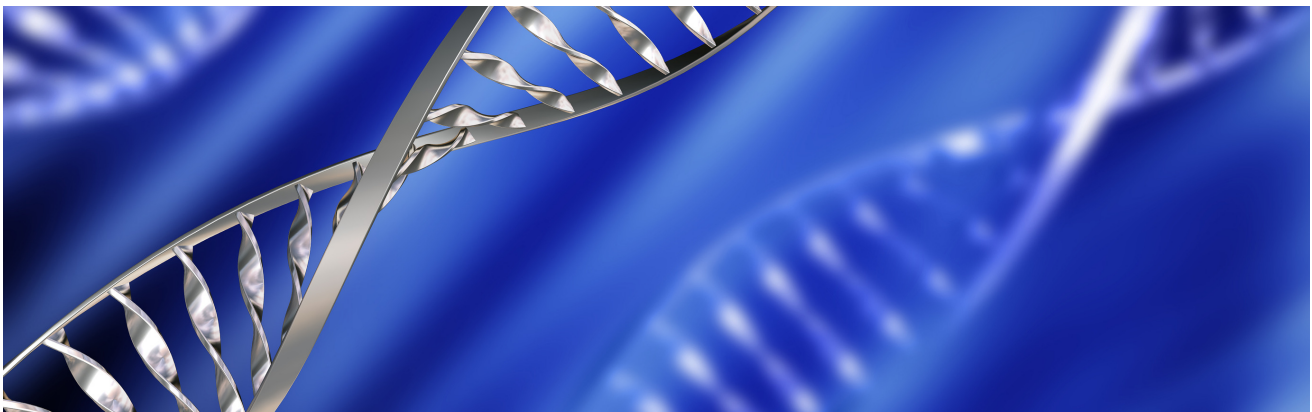
1250 Moursund Avenue, Houston, Texas 77030

Please contact Ashley LeMaire (832-331-4166) or Mark LeMaire (228-596-4026) for directions or assistance if you get lost.

Conference Schedule

Thursday

9:30 am	Childcare Drop-off & Registration Begins	
10:00 am	Welcome & Announcements	Kelly Mastin
10:15 am	Discovery & Benefits of Rare Disease Research	Hsiao-Tuan Chao, MD & Michael Wangler, MD
11:00 am	Genetics 101	Pilar L. Magoulas, MS, CGC
11:50 am	Lunch (60 min)	
12:55 pm	The Neurological Impact of HADDS	Hsiao-Tuan Chao, MD
1:40 pm	Clinical Study: HADDS & the NRI	Hsiao-Tuan Chao, MD
2:15 pm	Research Screenings until 5 pm	
2:15 pm	Mom & Dad Circles (90 min)	
5:30 pm	Informal Social at the Hilton Pool Area	



Conference Schedule

Friday

- Childcare on-site in NRI conference room on first floor during tour
- All Day Scheduled Clinic Visits with Dr. Chao & Dr. Wangler
- 9:30-11:30 am Jan & Dan Duncan Neurological Research Institute Tours (30 min each, adults only).
- 1:00-2:30 pm Jan & Dan Duncan Neurological Research Institute Tours (30 min each, adults only)
- 5:00 Informal Social at the Hilton Pool Area

Saturday

- 8:30 am Childcare Drop-off Begins
- 9:00 am What is HADDs? Non-Neurological and Genetic Findings Michael Wangler, MD
- 9:45 pm Potential Urological Implications of EBF3 Gene Mutation Irina Stanasel, MD
- 10:30 am Break
- 10:50 am Advocacy and Q & A Geraldine Bliss
- 11:40 am Lunch & EBF3 HADDs Foundation presentation Mark LeMaire & Mallory Schindler
- 12:45 pm IDEA: What You Need to Know Kelly Mastin
- 1:50 pm Future Directions Hsiao-Tuan Chao, MD

Speaker Bios



Hsiao-Tuan Chao, M.D., PH.D.

Assistant Professor of Molecular and Human Genetics,
Pediatrics, and Neuroscience
Baylor College of Medicine
Investigator, Jan and Dan Duncan Neurological
Research Institute, Texas Children's Hospital

Dr. Tuan Chao is a member of the faculty at the Jan and Dan Duncan Neurological Research Institute (NRI) at Texas Children's Hospital (TCH) and Baylor College of Medicine (BCM). She received her M.D. and Ph.D. degrees from BCM as a Medical Scientist Training Program McNair Scholar, supported by the McNair Medical Institute of The Robert and Janice McNair Foundation. She obtained her Ph.D. in Neuroscience from the laboratories of Dr. Huda Y. Zoghbi and Dr. Christian Rosenmund, where she explored the role of excitatory and inhibitory neuronal dysfunction in Rett syndrome. Following her doctoral training, Dr. Chao completed a child neurology residency at BCM and TCH. During her residency, she pursued postdoctoral training in the laboratory of Dr. Hugo J. Bellen, where she studied transcriptional dysregulation in neurodevelopmental disorders. Her postdoctoral work led to the recognition of the EBF3-related Hypotonia, Ataxia, and Delayed Development Syndrome.

Speaker Bios



Michael Wangler, M.D.

Assistant Professor

Department of Molecular and Human Genetics
Baylor College of Medicine

Michael Wangler completed his M.D. at Baylor College of Medicine in 2006, and completed Pediatric Residency at Texas Children's Hospital in 2009. He is board certified in Pediatrics and Medical Genetics. He is an Assistant Professor and physician scientist at the Jan and Dan Duncan Neurological Research Institute and in the Department of Molecular Human Genetics at Baylor College of Medicine. His research interests include rare and undiagnosed disease, and using genomics in medicine. He is an expert in HADDs syndrome as well as peroxisomal disorders, Xia-Gibbs syndrome, and genetic brain disorders.



Irina Stanasel, M.D.

Pediatric Urologist

Children's HealthSM

Assistant professor in

the Department of Urology at UT Southwestern Medical School

Irina Stanasel provides care for children of all ages who have a variety of urologic disorders. She received her medical degree from Baylor College of Medicine in Houston, Texas and completed her residency in urology at Wake Forest University Baptist Medical Center. She completed a pediatric urology fellowship at Texas Children's Hospital in Houston. Her specialties include

Speaker Bios

fetal urology, bladder bowel dysfunction, complex hypospadias repair, prune belly syndrome and quality improvement in pediatric urology. In addition to her medical degree, Dr. Stanasel holds a chemical engineering degree from the University of Texas, Austin. She also spent two years as a faculty member at the University of New Mexico working with underserved populations.



Pilar L. Magoulas, MS, CGC

Certified Genetic Counselor

Chief, Division of Genetic Counseling

Assistant Professor, Department of Molecular and Human Genetics

Baylor College of Medicine / Texas Children's Hospital

Pilar Magoulas received her Bachelor of Science degree in Psychology from the University of Florida in 2001 and a Master of Science degree in Genetic Counseling from Northwestern University in 2003. She currently works as a pediatric genetic counselor at Texas Children's Hospital where she serves as the Manager of the Pediatric Genetics clinic, and Chief of the Division of Genetic Counseling. Pilar is a member of the National Society of Genetic Counselors and American College of Medical Genetics. She serves on the Board of directors for CFC International, support group for individuals with Cardio-facio-cutaneous syndrome, on the Scientific Advisory Council for the National Foundation for Ectodermal Dysplasias, and the RASopathies Network USA, and serves on the Program Committee for the American College of Medical Genetics and Genomics.

Speaker Bios



Geraldine Bliss, MS

Board Member

Phelan-McDermid Syndrome Foundation

Geraldine Bliss is the parent of a child with Phelan-McDermid Syndrome (PMS), caused by a partial deletion of the SHANK3 gene. Due to the severity and refractory nature of her son's epilepsy, she made a promise to him to do everything she could to help him get better, which led her to become the research support committee chairperson of the Phelan-McDermid Syndrome Foundation (PMSF). She currently serves on PMSF's board of directors. Her vision and personal mission is to accelerate research that will lead to effective treatments and eventually cures for Phelan-McDermid Syndrome. Within the PMSF research portfolio is the Phelan-McDermid Syndrome International Registry, a web-based platform through which families upload genetic reports and share phenotypic information. In 2014, the Phelan-McDermid Syndrome Data Network, which merges registry data with electronic health data, became part of PCORnet.

Her other roles include the oversight of PMSF's grants and fellowship programs, the development of resources for research, such as biosample collections, the engagement of patients in clinical research studies and trials, patient education, and the organization of symposia and other scientific meetings.

Speaker Bios



Kelly Mastin

Board Member

EBF3 HADDS Foundation

Kelly is a mom raising three children, two of whom live life with a disability. Her youngest child, Chloe, has HADDS. Before becoming a mom, Kelly taught school at the high school and middle school level; this experience gives her a unique perspective as she advocates strongly for inclusion for her children. Kelly is a writer, advocate, and speaker with a passion for helping other parents who are on a similar journey. She currently serves on the board of the EBF3 HADDS Foundation.

Acknowledgements

Special thank you to the following organizations for supporting our conference!



©shellylambertphotography



Thank you to the many individuals and families near and far that volunteered their time and resources to help us organize this conference, including:

Siobhan Barney The Schindlers
Leslie Carroll Jay Richards

Thank you to Geraldine Bliss and her family for their support and mentoring as we have embarked on this rare disease advocacy journey.

Very special thank you to Pilar Magoulas for your support, guidance, planning, and organization for this event. We could not have done this without you!

Notes

Notes